



EDAR gene

ectodysplasin A receptor

Normal Function

The *EDAR* gene provides instructions for making a protein called the ectodysplasin A receptor. This protein is part of a signaling pathway that plays an important role in development before birth. Specifically, it is critical for interactions between two embryonic cell layers called the ectoderm and the mesoderm. In the early embryo, these cell layers form the basis for many of the body's organs and tissues. Ectoderm-mesoderm interactions are essential for the formation of several structures that arise from the ectoderm, including the skin, hair, nails, teeth, and sweat glands.

The ectodysplasin A receptor interacts with a protein called ectodysplasin A1 (produced from the *EDA* gene). On the cell surface, ectodysplasin A1 attaches to this receptor like a key in a lock. When these two proteins are connected, they trigger a series of chemical signals that affect cell activities such as division, growth, and maturation. Before birth, this signaling pathway controls the formation of ectodermal structures such as hair follicles, sweat glands, and teeth.

Health Conditions Related to Genetic Changes

hypohidrotic ectodermal dysplasia

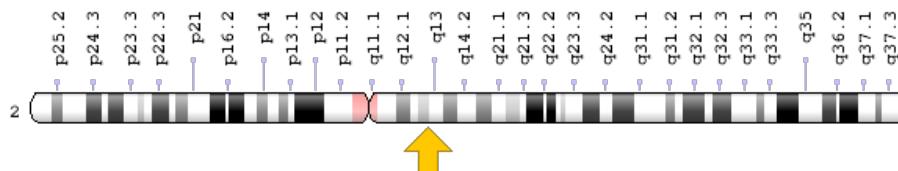
About 20 mutations in the *EDAR* gene have been identified in people with hypohidrotic ectodermal dysplasia. Most of these mutations change a single protein building block (amino acid) in the receptor protein, although deletions of genetic material from the *EDAR* gene also occur. Some *EDAR* mutations lead to the production of an abnormal version of the ectodysplasin A receptor. These genetic changes disrupt the signaling pathway needed for the formation of ectodermal structures such as hair follicles and sweat glands. When this type of mutation is present in one copy of the *EDAR* gene in each cell, it results in the autosomal dominant form of hypohidrotic ectodermal dysplasia.

Other *EDAR* mutations prevent cells from producing any ectodysplasin A receptor protein. As a result, the receptor is not available to trigger chemical signals that are necessary for ectoderm-mesoderm interactions and the normal development of ectodermal structures. This type of mutation, when present in two copies of the *EDAR* gene in each cell, causes an autosomal recessive form of hypohidrotic ectodermal dysplasia.

Chromosomal Location

Cytogenetic Location: 2q13, which is the long (q) arm of chromosome 2 at position 13

Molecular Location: base pairs 108,894,471 to 108,989,372 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DL
- ectodysplasin 1, anhidrotic receptor
- ectodysplasin A1 isoform receptor
- ectodysplasin receptor
- ED1R
- ED3
- ED5
- EDA-A1R
- EDA1R
- EDA3
- EDAR_HUMAN

Additional Information & Resources

Educational Resources

- Eda/Edar Signaling (Eurekah Bioscience Collection)
<https://www.ncbi.nlm.nih.gov/books/NBK6103/>

GeneReviews

- Hypohidrotic Ectodermal Dysplasia
<https://www.ncbi.nlm.nih.gov/books/NBK1112>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28EDAR%5BTIAB%5D%29+OR+%28ectodysplasin+A+receptor%5BTIAB%5D%29%29+OR+%28%28ectodysplasin+1,+anhidrotic+receptor%5BTIAB%5D%29+OR+%28ectodysplasin+A1+isoform+receptor%5BTIAB%5D%29+OR+%28ectodysplasin+receptor%5BTIAB%5D%29+OR+%28ED1R%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- ECTODYSPASIN A RECEPTOR
<http://omim.org/entry/604095>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_EDAR.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=EDAR%5Bgene%5D>
- HGNC Gene Family: Tumor necrosis factor receptor superfamily
<http://www.genenames.org/cgi-bin/genefamilies/set/782>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2895
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/10913>
- UniProt
<http://www.uniprot.org/uniprot/Q9UNE0>

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